

## Emma's Story

### A Grandmother's Quest

HB 5527

Everyone deserves a lifetime.

Thank you for allowing us to share our story of Emma Rilee Daniels.

My granddaughter **Emma Rilee Daniels** was born July 20, 2009, healthy and beautiful. She was my first grandchild. My son cried with *pride and joy* while welcoming his new beautiful little girl into our world.

Life was perfect for this new little family. My son had it all, a great career in the US Air Force, followed by a successful career as an Executive Chef in Princeton, NJ, a loving wife, and a perfectly healthy little girl. Emma was the center of Ryan and Nichole's world; nothing else existed or mattered. She met all her developmental milestones, smiled, and cooed. She had a voracious appetite and loved watching the colors on TV. She loved to babble to their golden retriever, Connor.

However, at 2 1/2 months old, this perfect little girl became fussy and irritable. We did not notice then, but looking back at photos, we see her thumbs are folded into her palms. She stopped smiling and cooing; her cry changed – it became a piercing scream. After several visits to the pediatrician we were told a series of diagnoses: colic, teething; irritable baby.

Sadly, I now know these are all classic symptoms of Krabbe (crab ā) disease, or Globoid Cell Leukodystrophy, an inherited neurodegenerative lysosomal enzyme disorder affecting the central and peripheral nervous systems. ***If testing for Krabbe had been included during the many tests already screened for Emma at birth would have had the opportunity at life.***

Instead, on 12/15/09, Emma was admitted to St. Peter's University Hospital. The doctors ran a CT scan – normal; her CBCs – normal; we were told neurological issues were not on the table. Because she continually cried, they moved on to metabolic testing and then discharged her saying we *may* want to get an EEG. Emma's mom took her to another pediatrician who immediately sent her to Children's Hospital of Philadelphia (CHOP). ***Had she been diagnosed in time, treatment could have ensued and Emma would have had the opportunity at life.***

Instead, at CHOP, the staff ran an MRI which indicated white matter involvement. They were suspicious of Krabbe but we were told the blood tests would take weeks to months to confirm an official diagnosis.

Because my daughter, Kate, works in the medical field, she had access to information that allowed us to do our own research. She connected with an organization, Hunter's Hope, founded by Jim Kelly. Their CEO, Jacque Waggoner, directed us to doctors at Duke Medical Center. Jacque explained to us the urgency of a diagnosis because every ticking minute was critical for any chance of intervention.

Dr Joanne Kurtzberg, Director of the Pediatric Blood and Marrow Transplant Program at Duke University Medical Center, made arrangements for Emma to arrive at their site in two days. By the time we arrived Dr. Kurtzberg had the official diagnosis from Dr. Wenger at Thomas Jefferson University in Phila. The diagnosis was achieved within 1 week, not the weeks or months we were told we had to wait.

It was Krabbe.

Devastation – The dreams and aspirations Emma's parents had for her wonderful, precious life turned to despair, sorrow, and the most profound sadness I have ever witnessed in my lifetime.

After Dr. Kurtzberg examined Emma she determined that our baby was no longer a candidate for any treatment; the only medical intervention available was to try to alleviate her pain.

On her six-month birthday, she had a feeding tube surgically implanted in her tiny little body. She lost her ability to suck and swallow. She was in chronic pain and scared. She had seizures. To watch a beautiful thriving little girl diminish in the way she did is a horror no parent or grandparent should have to live through.

Emma's fate was determined, she lived 2 ½ years in pain, medicated and then died. ***The next baby with this genetic disorder born in CT will go through the same thing. Instead, if broadened newborn screening, to include Krabbe, is approved as a state mandate, the next baby born with LSD can be saved.*** That is a fact. If Emma had been screened for lysosomal storage diseases (LSD's), she could have had a lifetime. Her smile would not be taken.

There is an even sadder fact. If Emma had been born just a few miles north over the border in New York State, she would have been tested for this disease at birth. She would then have been diagnosed and received treatment immediately to prevent the onset of the disease. Why should it matter what state a child is born in to receive benefit of the most current medical care?

Let's talk stats. Lysosomal storage disorders have been recognized as one of the major groups of genetic disorders affecting children. The rate of incidence for all LSDs in the human population is 1 in 5,000. With over 40 different disorders and a combined prevalence of up to one in 5,000 births, this group of disorders is a major public health problem and places an enormous burden, both financial and emotional, on affected individuals and their families as well as the public and private health systems. . 1 baby in 100,000 births will be born with Krabbe with or without screening. Early identification and diagnosis is essential since the most serious and debilitating symptoms – particularly neurological and skeletal – often do not respond to therapy once symptoms are present.

Currently, 5 of these disorders have validated newborn screening tests. Those disorders are Gaucher, Krabbe, Pompe, Niemann-Pick, and Fabry.

The Institute of Medicine's Committee states newborn screening only takes place 1) for conditions for which there are indications of clear benefit to the newborn, 2) when a system is in place for confirmatory diagnosis, and 3) when treatment and follow-up are available for affected newborns. Krabbe is treated by non-related cord blood transplant. While the treatment continues to improve, it is very dangerous, but I know Emma's parents would rather have held onto some hope of treatment versus the cold hard truth of just watching their baby being robbed of every ounce of breath.

Ethics aside, economics show that it is a cost benefit to the State of CT to screen and treat these children than to care for them while waiting for them to die.

Experience shows direct medical care for a child diagnosed with Krabbe costs \$700,000 annually. In cases where health care is not available, the state would be picking up these expenses

This translates to a 64% economic saving to CT if screening took place at birth and administered. Please see the enclosed data to understand the breakdown of expenses.

Illinois, Missouri, New Mexico, New York, New Jersey, and Pennsylvania have recently passed legislation to begin screening for these LSDs. Screening eliminates guessing and precludes misdiagnosis. In addition legislation is currently being heard in New Hampshire, Ohio, Maine, Wisconsin, Mississippi, Massachusetts, Tennessee, Kentucky, and Connecticut to expand newborn screening to include rare diseases.

As a result of Emma's Law in NJ and Hannah's Law in PA passing expanded newborn screening, the Children's Hospital of Philadelphia is creating a Leukodystrophy Center of Excellence for those babies who have been identified via newborn screening. Included in their multidisciplinary efforts, clinicians and researchers will be pushing the boundaries to fast-track enzyme and gene therapies that we believe will provide a cure within the next decade. The Leukodystrophy Center of Excellence at CHOP will assist those children diagnosed in time, to receive lifesaving cord blood transplants from our partners at Duke University and provide long-term family support and care through The Children's Hospital of Philadelphia's Integrated Care Services and Pediatric Advanced Care Team.

This is an extraordinary moment, something those of us who have personally witnessed the cruelty of leukodystrophy cannot believe has been achieved. What a great leap forward; to go from utter despair, to so much hope and optimism in just a few short years!

The real truth is without newborn screening there are no answers – we watched this little girl struggle to breathe, watched her seize, and watched her disappear before our eyes.

***You have the ability to make a difference to prevent the next baby born with Krabbe's from living this nightmare and offering parents and grandparents hope.***

The hope for these babies and their families is newborn screening. I humbly thank you for the opportunity share the story of an extraordinary, exquisite young lady, Emma Rilee Daniels.

Respectfully,

Sherri Daniels – Emma's Grandmother

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## **Newborn Screening is a Bargain**

Connecticut – 2009 live births –38,896 (2009 are the latest stats reported by CT that can be accurately compared to Medicaid stats) <http://www.statehealthfacts.org/profileind.jsp?ind=36&cat=2&rgn=8>

1:5000 diagnosed with LSD = 8 potential cases

\$700,000 annually for medical expenses to care for a child with LSD ([www.huntershope.org](http://www.huntershope.org))

$\$700,000 \times 8 = \$5,600,000$  (annual \$ care for children born w/ LSD)

$\$5,600,000 \times 2 = \$11,200,000$  – (\$ care for LDS children with expected lifespan of 2 years

# of CT Births financed by Medicaid = 14,500 (<http://www.statehealthfacts.org/comparemaptable.jsp?ind=222&cat=4>)

Potential of 3 children born annually with LSD under Medicaid (14,500/5000)

\$2,100,000 for 1 year of medical care for children on Medicaid

\$4,200,000 for 2 years of medical care for children on Medicaid

Transplant expense ranges from \$300,000 - \$700,000 (a one-time expense)  $\$500,000 \times 3 = \$1,500,000$

**64% Economic benefit is to screen at birth and administer treatment than to care for undiagnosed Medicaid cases**

**\$4,200,000 care for 2 years vs \$1,500,000 for treatment**

**Because of the rareness of these diseases, diagnosis is made too late for treatment to be an option. Newborn Screening is the only way for medical professionals to detect these disease in time for treatment to be a viable option.**